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Evolution HEALTHCARE

# Understanding and managing neuro-maturational differences * in primary care

## *ADHD, ASD, SLD, DCD

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# Overview

- **Conceptual Framework for Neuromaturational Differences**
- **Epidemiology**
- **Diagnostic Categories / Criteria**
- **Behavioural and  Educational Management**
- **Pharmacological Management**
- **Questions**

# Conceptual Framework for Neuromaturational Differences

# Concept 1:
# Deficits are in a continuum with the normal range

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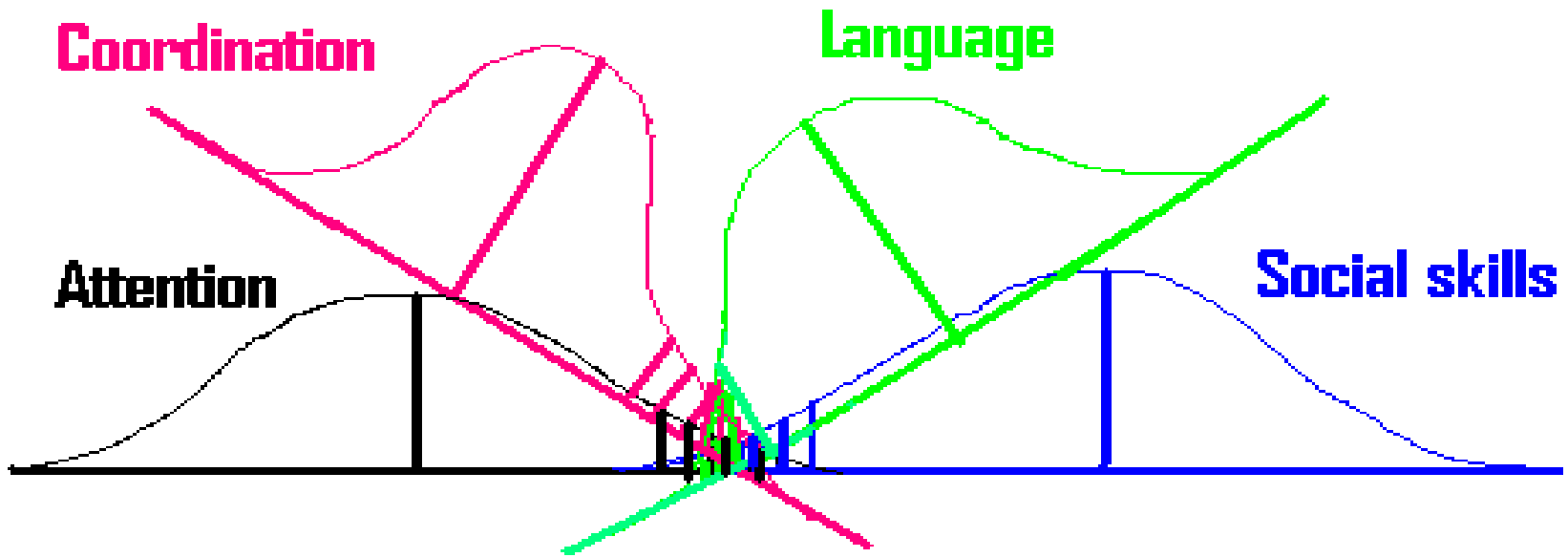
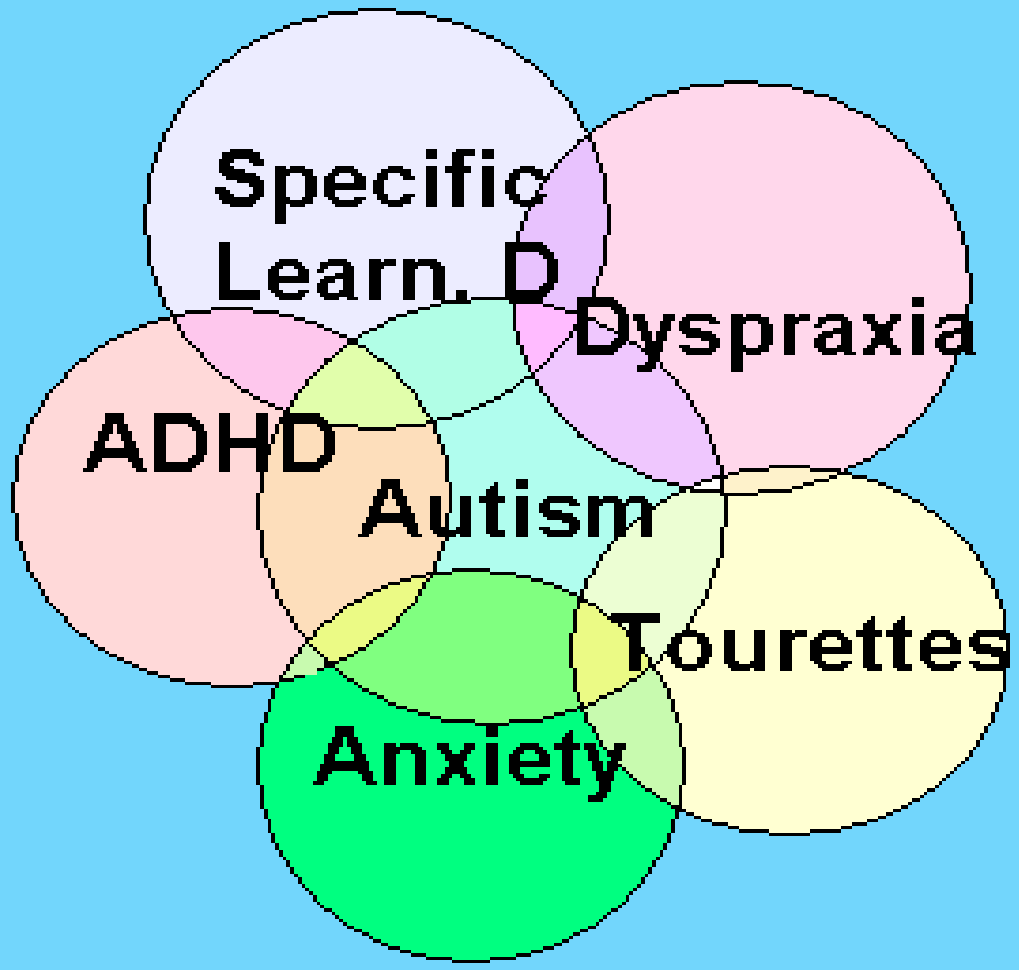
**Medical model diagnoses "disorder" for those outside typical range (2 SD) – better viewed as differences**



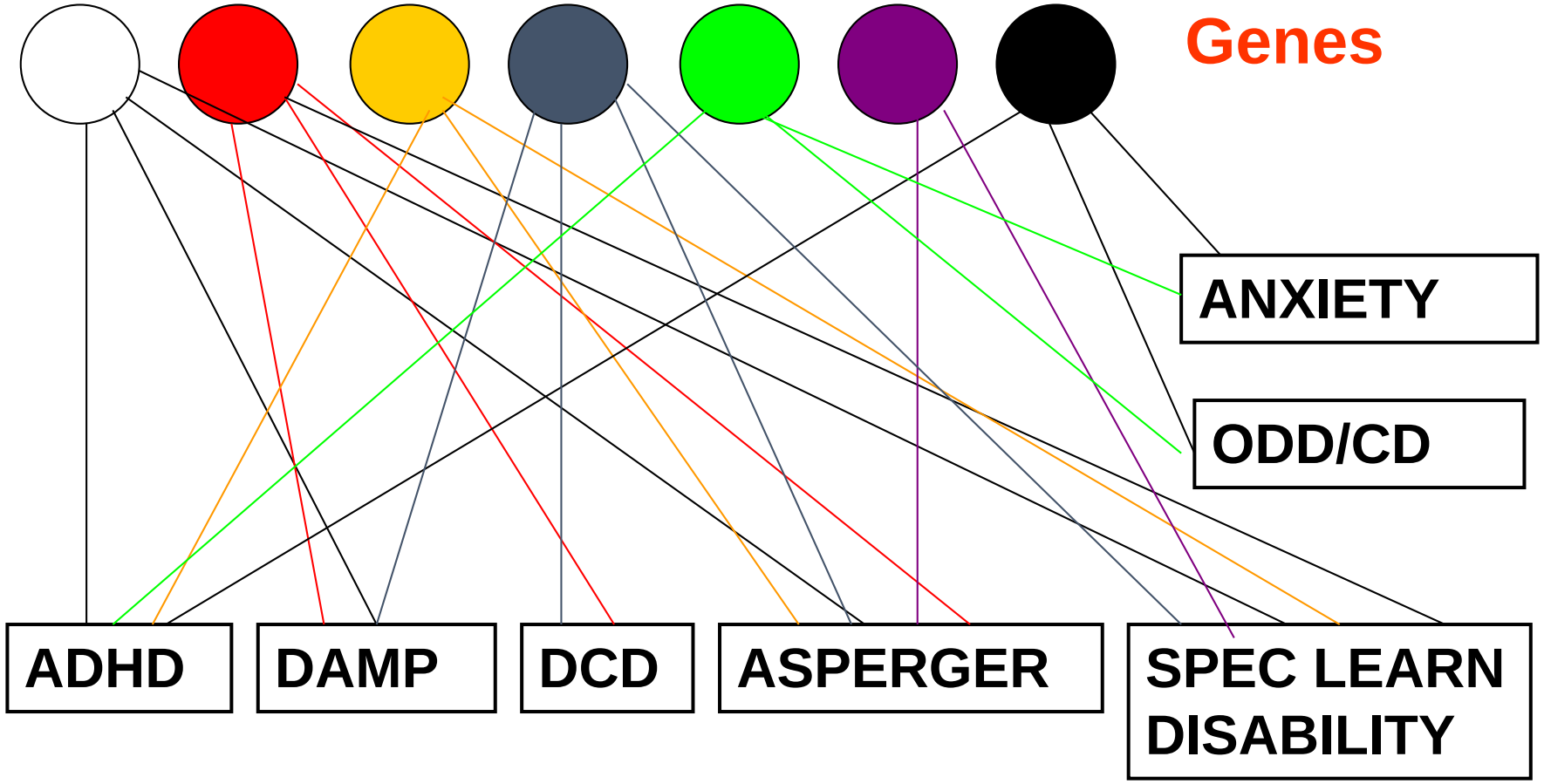
# Concept 2:
# Neuromaturational Difficulties Overlap




***Clumsiness, attention deficits, hyperactivity, social difficulties, anxiety and learning disabilities often cluster together***

***Always consider the Environment (Abuse / Neglect / Parental Mental Health) as potent contributors or causes of all these symptoms***

# Concept 3: Cause is largely polygenic



# Concept 3: Cause is largely polygenic



- **All these conditions share genetic predisposition;**
- **Autism twin studies 70-80% genetic**
  - 50% monozygotic concordance, 5% dyzygotic concordance
  - Siblings 50-fold risk compared to population risk
- **Multiple interacting genes**
- **Family history invariably present**

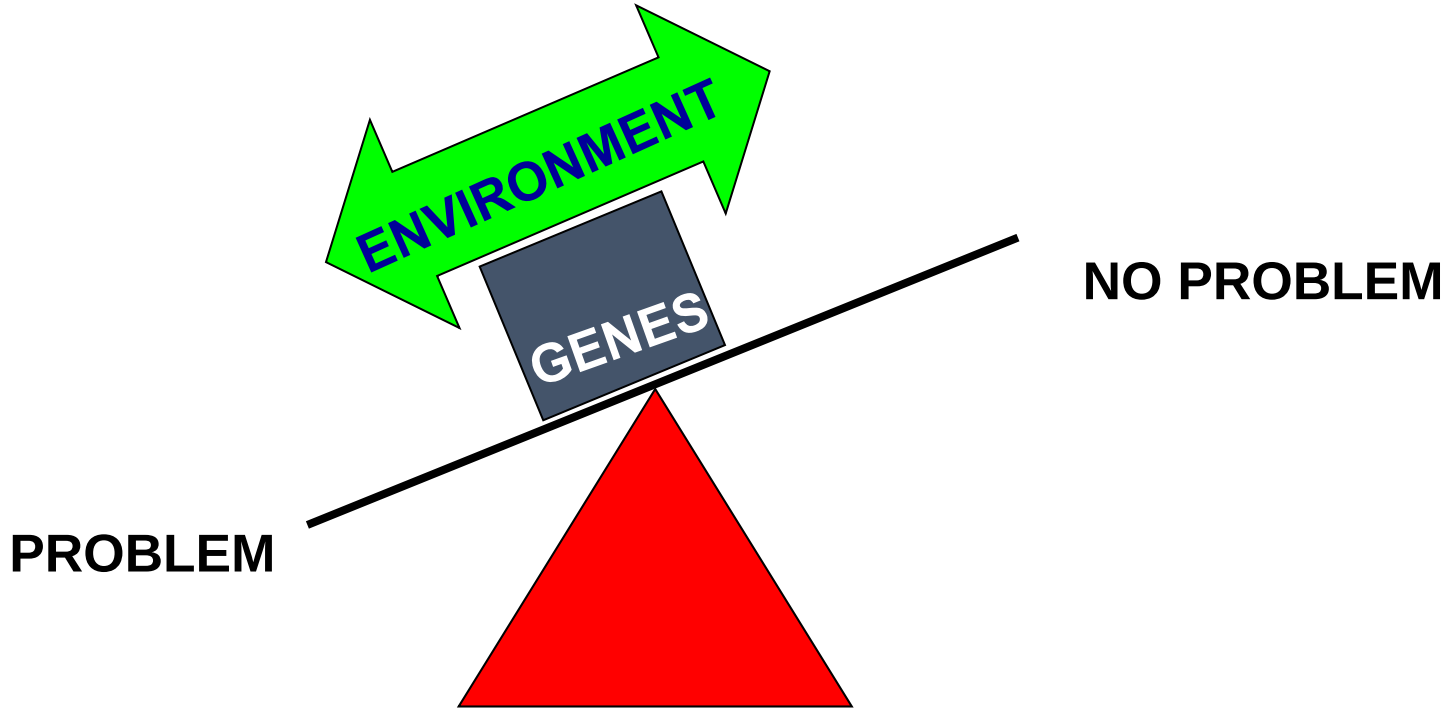
**In 2001, 4.7 % of the workforce and 3.65 % of the male NZ European workforce was employed in the IT industry**

**In children with Autism, 8/26 = 31% Dads worked in IT and telecommunications *(in 2 yr local cohort in 2003-04 of 68)***

***Evidence that engineering, science and accountancy are over-represented in fathers of children with Autism***

***Evidence of a 'peak and trough" cognitive profile in Autism relatives (better rote-learned and spatial abilities and difficulties with executive function)***

# Environment/Gene Interaction



# Disability or Difference?

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## Social Model of Disability:

**An impairment is only disabling if the environment is not adapted to it (WHO)**

- "wiring difference"
- Patterns of strengths as well as weaknesses
- Cultural Identity
- Evolutionary perspective

# Epidemiology

# Epidemiology



- Previously quoted: Autism = 5 per 10 000
- Recent studies 1 per 1000*
- Autistic Spectrum Disorders 3-4 per 1000 (some estimates go up to 1:100 for ASD traits)
- Male:female 4:1
- Prevalence stable ? (apparent or real increase?)

* 21800 Japanese children prospective to 3 years 1.3 Autism + 0.7 Autistic Traits Questionnaire all children 6-14 Nova Scotia (20800) 1 per 1000

* Sweden 3-17yr olds 1-2 per1000 Autism, 3 per 1000 Aspergers

# Why the increase ?

- Changes in diagnostic criteria over time
- Differences in methods used in studies
- Increasing awareness amongst professionals and the wider community
- Recognition that ASD:
  - occurs in association with other conditions (eg. ID, physical disability, syndromes, psychiatric conditions)
  - could occur in people with high IQ
  - presentation can be subtle.
- The question as to whether there has been a genuine increase remains open
- Evidence for higher rates in older fathers

# Environmental Triggers?



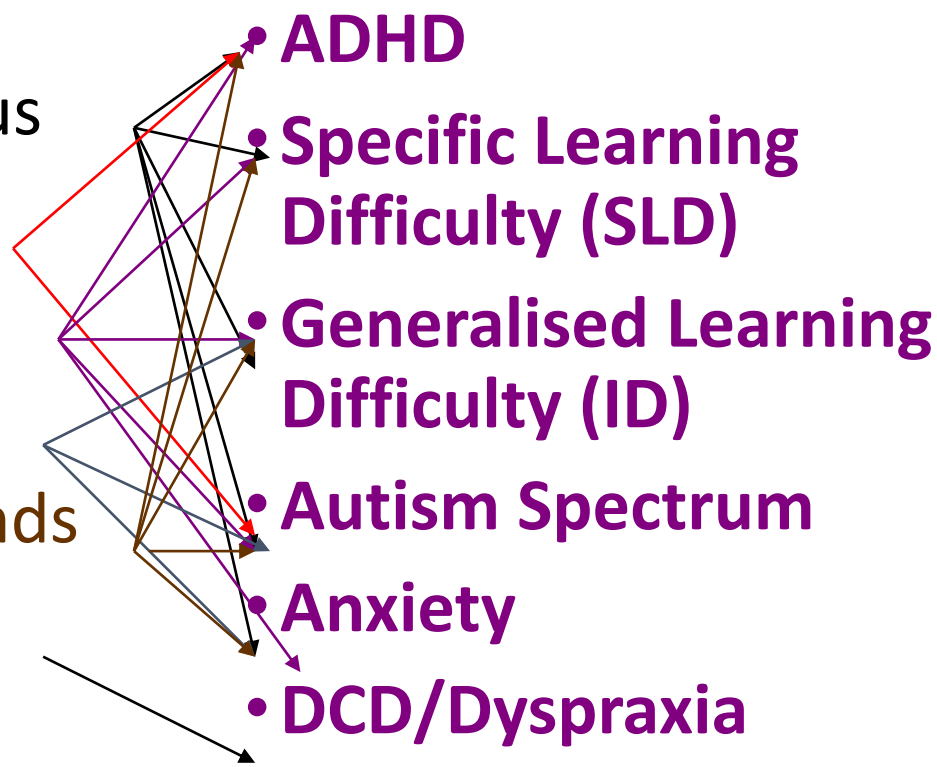
- Not MMR
- Leaky gut → dietary peptides acting as neurotransmitters?
- Viral infections
- In utero infections
- Maternal distress in pregnancy
- Congenital Measles/Rubella
- Metabolic conditions: untreated  PKU

# Diagnostic Categories / Criteria

# GP diagnosis

## What are the major problems at school and home?

- Concentration / focus
- Hyperactivity
- Behind in learning
- Tearful / no friends
- Aggressive / no friends
- Clumsy



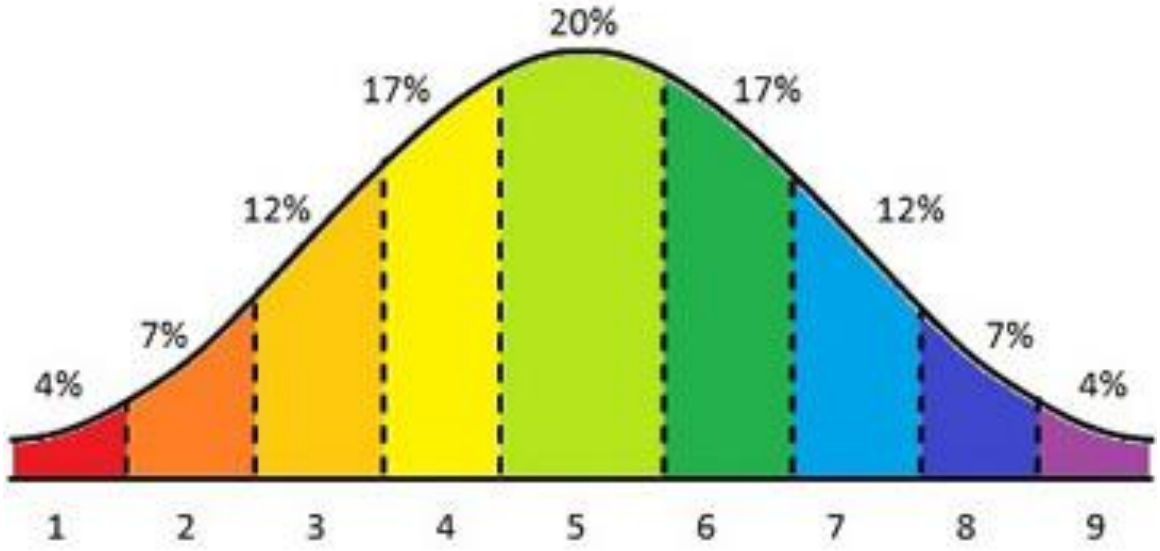
- **ADHD**
- **Specific Learning Difficulty (SLD)**
- **Generalised Learning Difficulty (ID)**
- **Autism Spectrum**
- **Anxiety**
- **DCD/Dyspraxia**

# Considerations in sorting out diagnosis



- **Have these problems always been there or are they new? (think anxiety, home environment, bullying, abuse)**
- **Are these problems very different at home compared to school (think environment)**
- **Is there a difference between achievement level and ability level? (ADHD / behaviour, environment, specific learning difficulty, hearing/vision)**
- **Need feedback from teacher directly (letter, email, phone-call, +/- standardized tools (Connors etc)**
- **Teacher needs if possible to give age-level for achievement in curriculum areas, or stanines, or both**

# Stanines and achievement levels




Developmental age
---------- x 100 = developmental quotient
Chronological age

An 8 year old working at a 6 year old level has a “DQ” of 75 which is around 2 SD from the mean, close to diagnostic level for Intellectual disability (IQ = 70)

## Unifying concepts in developmental disorders:

- **Significantly different what would be expected of a child their age.**
- **Present since early childhood**
- **Pervasive (observed in multiple settings)**
- **Causing impairment**

# Referral for diagnostic assessment



- Developmental Paediatric, MDT or General Paediatric Assessment
  - Diagnose underlying medical or developmental problem (Consider CGH microarray, MRI, EEG, Fragile X, hearing tests as indicated)
- Range of assessment tests:
  - Ability Tests Bayleys or Griffiths Developmental assessment or for older children Apititude/IQ tests such as WISC (“how the wiring works”)
  - Achievement tests (reading, spelling, writing such as Neale, WRAT)
  - Diagnostic tests (specific functions such as Reynell for language comprehension, SCAN for Auditory Processing Disorder, ADOS for Autism)
- Accurate diagnosis
- Child-centered plan
  - Generated and followed by parents, school, other professionals
- Follow-up
  - Follow progress, audit goals, and reformulate plan as required

# Global Developmental Disability vs Intellectual Disability by DSM 5



**GDD**

- Impairment in "*2 or more* domains of development"
- Diagnostic and predictive accuracy increases if utilize a higher threshold (deficits in all domains)

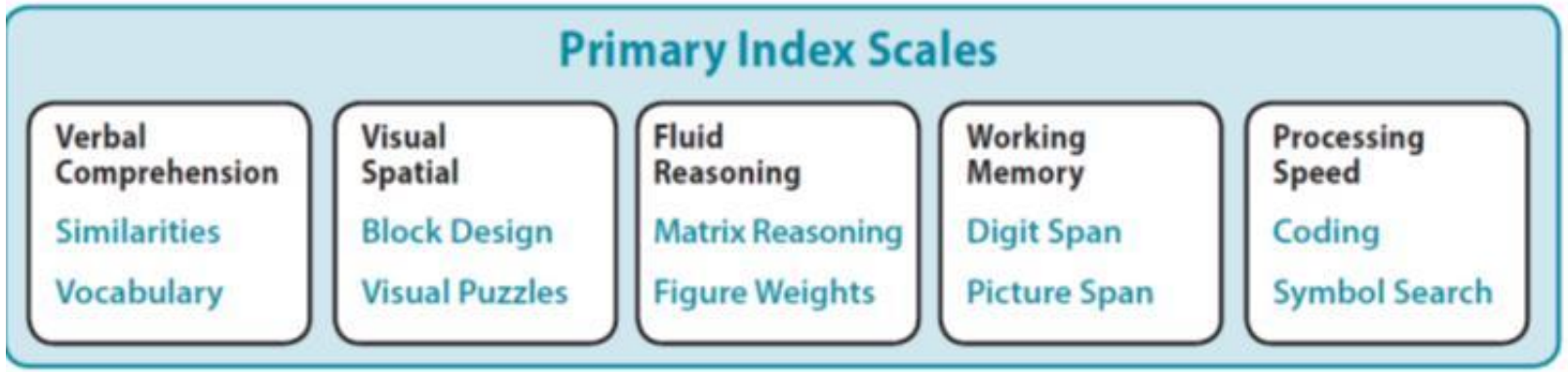
**ID**

1. Deficits in **intellectual functioning**
   **Reasoning;** **Abstract thinking;**
   **Problem solving;** **Judgment;**
   **Planning;** **Academic learning;**
   **Experiential learning**
   These mental abilities are measured by IQ tests. A score of approximately two standard deviations below average represents a significant cognitive deficit. This is typically an IQ score of 70 or below.
2. Deficits or impairments in **adaptive functioning**
   - **Communication:**
   - **Social skills:**
   - **Personal independence at home or in community settings:**
   - **School or work functioning:**
3. These limitations occur during the developmental period; were evident during childhood or adolescence

# Measuring Intelligence using WISC V



- **Verbal Comprehension**
  - expressive / receptive language
- **Performance Skills**
  - visuo-spatial perception
  - fine motor skills
  - construction skills
  - sequencing
- **Processing Speed**
- **Memory**
  - immediate / working / delayed
  - verbal / visual
  - recall / recognition
- **Attention**
  - auditory / visual
  - selective / sustained / switching
  - dual task
- **Social judgement**
- **Executive functions**
  - Planning
  - Problem solving
  - Use of feedback
  - Shifting
  - Flexibility
  - Monitoring

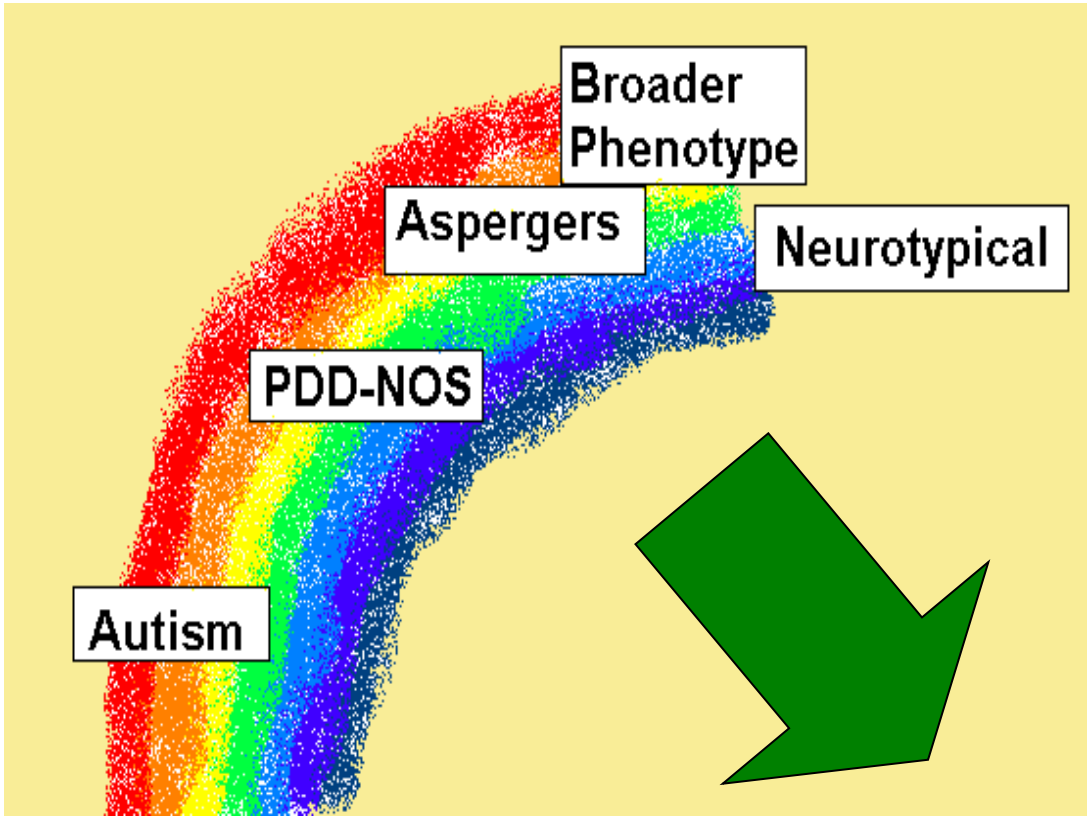


# Specific Learning Disabilities DSM 5



- Criterion A
  - at least one of six symptoms of learning difficulties that have persisted for at least 6 months despite the provision of extra help or targeted instruction).
- Criterion B
  - the affected academic skills are substantially and quantifiably below those expected for age and cause impairment in academic, occupational, or everyday activities, as confirmed by individually administered standardized achievement measures and comprehensive clinical assessment.
- Criterion C
  - onset during the school-age years, although may not fully manifest until young adulthood in some individuals
- Criterion D
  - specifies which disorders (Intellectual Disabilities, uncorrected auditory or visual acuity problems, other mental or neurological disorders) or adverse conditions (psychosocial adversity, lack of proficiency in the language of instruction, inadequate instruction) must be ruled out before a diagnosis of SLD can be confirmed.

# DSM 5




- Cultural identity of “Aspie” community removed by revision of DSM?
- Aspergers may still have relevance in terms of books/resources?

**ASD**

# DSM 5 Autism Spectrum Disorder
*Must meet criteria 1, 2, and 3:*

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1. Clinically significant, persistent deficits in social communication and interactions, as manifest by all the following:

a. Marked deficits in nonverbal and verbal communication and interaction

b. Marked deficits in social-emotional reciprocity

c. Failure to develop and maintain peer relationships appropriate to developmental level

2. Restricted, repetitive patterns of behavior, interests, and activities, as manifested by at least TWO of the following:

a. Stereotyped or repetitive speech, motor movements, or use of objects;

b. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behaviour, or excessive resistance to change;

c. Highly restricted, fixated interests that are abnormal in intensity or focus;

d. Hyper-or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment

# DSM 5 Autism Spectrum Disorder



3. Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities)

4. Symptoms together limit and impair everyday functioning

Further distinctions will be made according to severity levels.

The severity levels are based on the amount of support needed, due to challenges with social communication and restricted interests and repetitive behaviours. For example, a person might be diagnosed with ASD, Level 1, 2, or 3.

# DSM 5 ASD Severity levels

| ASD | 1 Mild | 2 Moderate | 3 Severe |
|---|---|---|---|
| **Social Communication Domain** | Without supports in place, deficits in social communication cause noticeable impairments. Has difficulty initiating social interactions and demonstrates clear examples of atypical / unsuccessful responses to social overtures of others. May appear to have decreased interest in social interaction | Marked deficits in verbal and nonverbal social communication skills; social impairments apparent even with supports in place; limited initiation of social interactions and reduced or abnormal response to social overtures from others | Severe deficits in verbal and nonverbal social communication skills cause severe impairments in functioning; very limited initiation of social interactions and minimal response to social overtures from others. |
| | | | |

# DSM 5 ASD Severity levels

| ASD | 1 Mild | 2 Moderate | 3 Severe |
|---|---|---|---|
| | | | |
| **ASD Behaviour domain** | Rituals and repetitive behaviours (RRB's) cause significant interference with functioning in one or more contexts. Resists attempts by others to interrupt RRB's or to be redirected from fixated interest. | RRBs and/or preoccupations or fixated interests appear frequently enough to be obvious to the casual observer and interfere with functioning in a variety of contexts. Distress or frustration is apparent when RRB's are interrupted; difficult to redirect from fixated interest. | Preoccupations, fixated rituals and/or repetitive behaviours markedly interfere with functioning in all spheres. Marked distress when rituals or routines are interrupted; very difficult to redirect from fixated interest or returns to it quickly. |

# DSM 5 ADHD Criteria



**Inattention:**

- **Six or more symptoms of inattention for children up to age 16, or five or more for adolescents 17 and older and adults;**
- **Symptoms of inattention have been present for at least 6 months, and they are inappropriate for developmental level:**
  - Often fails to give close attention to details or makes careless mistakes in schoolwork, at work, or with other activities.
  - Often has trouble holding attention on tasks or play activities.
  - Often does not seem to listen when spoken to directly.
  - Often does not follow through on instructions and fails to finish schoolwork, chores, or duties in the workplace (e.g., loses focus, side-tracked).
  - Often has trouble organizing tasks and activities.
  - Often avoids, dislikes, or is reluctant to do tasks that require mental effort over a long period of time (such as schoolwork or homework).
  - Often loses things necessary for tasks and activities (e.g. school materials, pencils, books, tools, wallets, keys, paperwork, eyeglasses, mobile telephones).
  - Is often easily distracted
  - Is often forgetful in daily activities.

# DSM 5 ADHD Criteria



**Hyperactivity and Impulsivity:**

- **Six or more symptoms of hyperactivity-impulsivity for children up to age 16, or five or more for adolescents 17 and older and adults;**
- **Symptoms of hyperactivity-impulsivity have been present for at least 6 months to an extent that is disruptive and inappropriate for the person's developmental level:**
  - Often fidgets with or taps hands or feet, or squirms in seat.
  - Often leaves seat in situations when remaining seated is expected.
  - Often runs about or climbs in situations where it is not appropriate (adolescents or adults may be limited to feeling restless).
  - Often unable to play or take part in leisure activities quietly.
  - Is often "on the go" acting as if "driven by a motor".
  - Often talks excessively.
  - Often blurts out an answer before a question has been completed.
  - Often has trouble waiting his/her turn.
  - Often interrupts or intrudes on others (e.g., butts into conversations or games)

# DSM 5 ADHD Criteria

**In addition, the following conditions must be met:**

- Several inattentive or hyperactive-impulsive symptoms were present before age 12 years.
- Several symptoms are present in two or more setting, (e.g., at home, school or work; with friends or relatives; in other activities).
- There is clear evidence that the symptoms interfere with, or reduce the quality of, social, school, or work functioning.
- The symptoms do not happen only during the course of schizophrenia or another psychotic disorder. The symptoms are not better explained by another mental disorder (e.g. Mood Disorder, Anxiety Disorder, Dissociative Disorder, or a Personality Disorder).

# Behavioural and Educational Management

# Medical Management

- Formal Audiology
- CGH microarray, Fragile X, TFTs, urine metabolic screen
- FBC and iron studies if restricted diet, lead if Pica, full metabolic workup if regression (except for isolated language regression)
- Child Development Team
  - VNDT and/or SLT and/or Psychologist and/or OT
- Special Education Early Intervention Team
  - Speech Language Therapist, Early intervention teacher, Psychologist
- NASC
  - Home Help and Respite Care
- Behaviour Management Service (Explore)
  - Behaviour management assessment, support and advice for children and adults with intellectual disability or autism
  - Parent training / education (ASD+, TIPS, Teenlife etc)
- WINZ
  - Child Disability Allowance
  - Disability Allowance (means tested)
  - Supported Living Payment
- Paediatric Follow-up
  - Advocacy, anticipatory Guidance, medications where required
- Service organisations (Autism NZ, Altogether Autism etc)
  - Parent support
  - Information sources

# Behavioural Management

Look for ABC's

- Antecedents
- Behaviour (specific)
- Consequences

FIDO

- Frequency
- Intensity
- Duration
- Outcome

# Behaviour Strategies

- Instructions
  - Brief, concise, concrete, direct
- Break things down into small steps
- Make it a discovery not advice
  - “what happens if....”
- Prepare for changes
  - “First.... Then...”
- Be specific when rewarding a child
  - “doing.... was the right thing to do”
- Social stories
  - www.thegraycenter.org

- Environment
  - Predictable / Consistent / Calm
- Visual communication
  - Visual timer
  - Pictures “First... Then...”
  - Comic strip conversations
  - Signs
- Desensitisation
- Token systems
  - need to ensure reward is reinforcing

# Difficult behaviour in Autism:



## Causes:

Anxiety – often social anxiety

Communication

Sensory (seeking/avoiding/ overload)

“Hard-wired” / intrinsic

## Consequences:

Obsessive repetitive behaviour

Aggression and self harm

Irritability

Hyperactivity

Social withdrawal or phobias

Medication trials can only be planned and managed effectively if the cause of the target behaviour is known

# Behavioural management ASD

- Consistent routine
- Visual prompts to aid understanding of expectations, transitions and sequences
- Modification of environment to compensate for sensory overload / overstimulation

# Management – Specific Learning Disability

- Organise complex tasks into simple steps and prompt
- Reduce distraction (sit at front next to studious kids)
- Encourage/reward achievement
- Allow self-regulation for stress (time-out area, run around, squeeze a ball)
- Lunchtime detention for slow work completion inhumane
- Private tutoring/SPELD (for bypass pathways or improve pathways, not “more of the same)

# Pharmacological Management

# Principles and Practice of Pharmacotherapy in developmental disabilities



- There is **no** medication to treat autism or intellectual disability, only medications to manage some symptoms/associated behaviours
- Environmental manipulation and behavioural / educational management **always** the first step and **always** used in partnership with medications
- Start low, go slow, one change at a time, monitor response
- Medication trials with behavioural targets have high placebo response
- Plan to withdraw medication in future to assess continued benefits and side effects.
- Everything with benefits has risk
- These medications are evidence-based, but used ‘off label”
- Use the internet cautiously

| | |
|---|---|
| Stimulants<br>Methylphenidate<br>Dexamphetamine | Hyperactive, impulsive behaviour and short attention span |
| Clonidine/Atomoxetine | Mixed profile |
| Atypical Antipsychotics<br>Risperidone<br>Aripiprazole | Aggressive, disturbed, anxious behaviour |
| SSRI antidepressants<br>Fluoxetine<br>Sertraline<br>Citalopram | Obsessive compulsive, anxious behaviour |

Melatonin

Sleep

Omega 3

General brain enhancement?

Anticonvulsants

Epilepsy (occasionally behaviour)

Nutritional support

- Vitamins
- Minerals
- Restrictions

# In Summary

# Summary

- Developmental disabilities present with developmental and behavioural features which are significantly different to what would be expected of a child their age.
- Features have been present since early childhood (but may manifest or cause more impairment as the child gets older)
- They are pervasive (observed in multiple settings)
- They cause significant impairment in function
- They present as patterns of strengths and weaknesses, and can result in significant emotional and financial strain for the whanau
- Awareness of the genetic and environmental contributors allows earlier recognition of risk factors
- The earlier the diagnosis is made and appropriate interventions are put in place the better the outcome

For more information
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